platyspondylic lethal skeletal dysplasia, Torrance type

Platyspondylic lethal skeletal dysplasia, Torrance type is a severe disorder of bone growth. People with this condition have very short arms and legs, underdeveloped pelvic bones, and unusually short fingers and toes (brachydactyly). This disorder is also characterized by flattened spinal bones (platyspondyly) and an exaggerated curvature of the lower back (lordosis). Infants with this condition are born with a small chest with short ribs that can restrict the growth and expansion of the lungs.

As a result of these serious health problems, some affected fetuses do not survive to term. Infants born with platyspondylic lethal skeletal dysplasia, Torrance type usually die at birth or shortly thereafter from respiratory failure. A few affected people with milder signs and symptoms have lived into adulthood.

Frequency

This condition is very rare; only a few affected individuals have been reported worldwide.

Genetic Changes

Platyspondylic lethal skeletal dysplasia, Torrance type is one of a spectrum of skeletal disorders caused by mutations in the *COL2A1* gene. This gene provides instructions for making a protein that forms type II collagen. This type of collagen is found mostly in the clear gel that fills the eyeball (the vitreous) and in cartilage. Cartilage is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears. Type II collagen is essential for the normal development of bones and other connective tissues that form the body's supportive framework.

All of the *COL2A1* mutations that have been found to cause platyspondylic lethal skeletal dysplasia, Torrance type occur in a region of the protein called the C-propeptide domain. These mutations interfere with the assembly of type II collagen molecules, reducing the amount of this type of collagen in the body. Instead of forming collagen molecules, the abnormal COL2A1 protein builds up in cartilage cells (chondrocytes). These changes disrupt the normal development of bones and other connective tissues, leading to the skeletal abnormalities characteristic of platyspondylic lethal skeletal dysplasia, Torrance type.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- platyspondylic chondrodysplasia, Torrance-Luton type
- platyspondylic skeletal dysplasia, Torrance type
- PLSD-T
- PLSD-TL

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Platyspondylic lethal skeletal dysplasia Torrance type
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1835437/

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Lordosis
  https://medlineplus.gov/ency/article/003278.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Lordosis
  https://medlineplus.gov/ency/article/003278.htm
- Health Topic: Bone Diseases
  https://medlineplus.gov/bonediseases.html

- Health Topic: Connective Tissue Disorders
  https://medlineplus.gov/connectivetissuedisorders.html
- Health Topic: Dwarfism
  https://medlineplus.gov/dwarfism.html

Genetic and Rare Diseases Information Center

- Platyspondylic lethal skeletal dysplasia Torrance type
  https://rarediseases.info.nih.gov/diseases/4382/platyspondylic-lethal-skeletal-dysplasia-torrance-type

Additional NIH Resources

- National Institute of Arthritis and Musculoskeletal and Skin Diseases
  https://www.niams.nih.gov/Health_Info/Connective_Tissue/

Educational Resources

- MalaCards: platyspondylic skeletal dysplasia, torrance type
  http://www.malacards.org/card/platyspondylic_skeletal_dysplasia_torrance_type
- Nemours Children's Health System: Skeletal Dysplasia
  https://www.nemours.org/service/medical/skeletal-dysplasia.html?tab=about
- Orphanet: Platyspondylic lethal chondrodysplasia
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1417

Patient Support and Advocacy Resources

- Human Growth Foundation
  http://hgfound.org/
- International Skeletal Dysplasia Registry, UCLA
  http://ortho.ucla.edu/isdr
- Little People of America
  http://www.lpaonline.org/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/dwarfism.html

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28platyspondylic+skeletal+dysplasia%5BTIAB%5D%29+OR+%28platyspondylic+lethal+skeletal+dysplasia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- PLATYSPONDYLIC LETHAL SKELETAL DYSPLASIA, TORRANCE TYPE
  http://omim.org/entry/151210

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12961049
- Nishimura G, Nakashima E, Mabuchi A, Shimamoto K, Shimamoto T, Shimao Y, Nagai T, Yamaguchi T, Kosaki R, Ohashi H, Makita Y, Ikegawa S. Identification of COL2A1 mutations in platyspondylic skeletal dysplasia, Torrance type. J Med Genet. 2004 Jan;41(1):75-9.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14729840
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1757240/
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